

A B S T R A C T

Title: Methods

This invention relates to polymorphisms in the human pyruvate dehydrogenase E1 β
5 (PDH E1 β) gene. The invention also relates to methods and materials for analysing allelic
variation in the PDH E1 β gene, and to the use of PDH E1 β polymorphism in the diagnosis
and treatment of diseases in which modulation of pyruvate dehydrogenase activity could be of
therapeutic benefit, such as diabetes, asthma, obesity, sepsis and peripheral vascular disease.
In particular, the invention is based on the discovery of a single nucleotide polymorphism in
10 the coding region of the human PDH E1 β gene, and three single nucleotide polymorphisms in
the 3' untranslated region (3'UTR) of the human PDH E1 β gene.